ACADS gene

acyl-CoA dehydrogenase, C-2 to C-3 short chain

Normal Function

The *ACADS* gene provides instructions for making an enzyme called short-chain acyl-CoA dehydrogenase (SCAD). This enzyme functions within mitochondria, the energy-producing centers within cells. SCAD is essential for fatty acid oxidation, which is the multistep process that breaks down (metabolizes) fats and converts them to energy.

SCAD is required to metabolize a group of fats called short-chain fatty acids. These fatty acids are found in some foods and are also produced when larger fatty acids are metabolized. Fatty acids are a major source of energy for the heart and muscles. During periods without food (fasting), fatty acids are also an important energy source for the liver and other tissues.

Health Conditions Related to Genetic Changes

short-chain acyl-CoA dehydrogenase deficiency

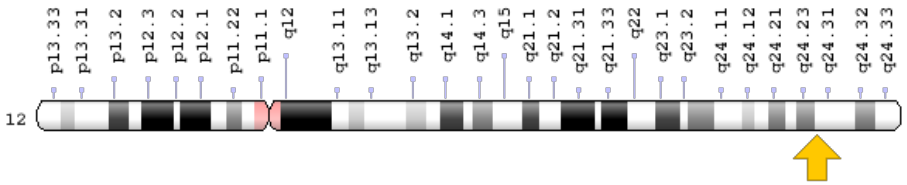
More than 55 mutations in the *ACADS* gene have been found to cause short-chain acyl-CoA dehydrogenase (SCAD) deficiency. Almost all of these mutations change single protein building blocks (amino acids) in the SCAD enzyme. These mutations prevent the enzyme from properly metabolizing short-chain fatty acids. As a result, these fats are not converted into energy, which can lead to the characteristic signs and symptoms of this disorder, including lack of energy (lethargy), low blood sugar (hypoglycemia), poor muscle tone (hypotonia), and weakness.

Researchers have also identified two common variations (polymorphisms) in the *ACADS* gene that each change one amino acid in the SCAD enzyme. Unlike other changes in the *ACADS* gene, these polymorphisms do not cause SCAD deficiency but may increase a person's risk of developing this disorder. One of these polymorphisms replaces the amino acid arginine with the amino acid tryptophan at protein position 147 (written as Arg147Trp or R147W). The other polymorphism switches the amino acid glycine with the amino acid serine at protein position 185 (written as Gly185Ser or G185S). Other genetic and environmental factors likely influence the risk of developing SCAD deficiency when a person carries either of these polymorphisms.

Chromosomal Location

Cytogenetic Location: 12q24.31, which is the long (q) arm of chromosome 12 at position 24.31

Molecular Location: base pairs 120,725,738 to 120,740,008 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ACAD3
- ACADS_HUMAN
- acyl-Coenzyme A dehydrogenase, C-2 to C-3 short chain precursor
- Butyryl-CoA dehydrogenase
- Butyryl dehydrogenase
- SCAD
- Unsaturated acyl-CoA reductase

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): The Utilization of Fatty Acids as Fuel Requires Three Stages of Processing
  https://www.ncbi.nlm.nih.gov/books/NBK22581/

GeneReviews

- Short-Chain Acyl-CoA Dehydrogenase Deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK63582

Genetic Testing Registry

- GTR: Genetic tests for ACADS
  https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=35%5Bgeneid%5D

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28ACADS%5BTIAB%5D%29+OR+%28short-chain+acyl-CoA+dehydrogenase%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- ACYL-CoA DEHYDROGENASE, SHORT-CHAIN
  http://omim.org/entry/606885

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ACADS%5Bgene%5D
- HGNC Gene Family: Acyl-CoA dehydrogenase family
  http://www.genenames.org/cgi-bin/genefamilies/set/974
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=90
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/35
- UniProt
  http://www.uniprot.org/uniprot/P16219

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